



MANBA gene

mannosidase beta

Normal Function

The *MANBA* gene provides instructions for making the enzyme beta-mannosidase. This enzyme works in the lysosomes, which are compartments that digest and recycle materials in the cell. Within lysosomes, the enzyme helps break down complexes of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins). Beta-mannosidase is involved in the last step of this process, helping to break down complexes of two sugar molecules (disaccharides) containing a sugar molecule called mannose.

Health Conditions Related to Genetic Changes

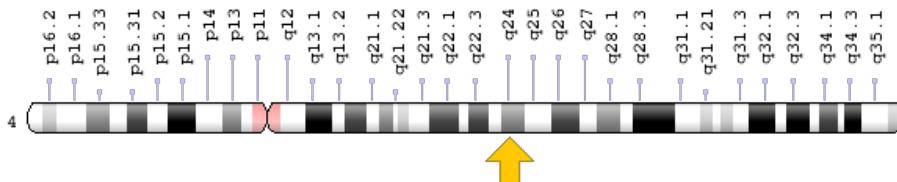
beta-mannosidosis

Approximately 12 mutations that cause beta-mannosidosis have been identified in the *MANBA* gene. The mutations result in a beta-mannosidase enzyme with little or no activity, and interfere with the ability of the enzyme to perform its role in breaking down mannose-containing disaccharides. These disaccharides gradually accumulate in the lysosomes and cause cells to malfunction, resulting in the signs and symptoms of beta-mannosidosis.

Chromosomal Location

Cytogenetic Location: 4q24, which is the long (q) arm of chromosome 4 at position 24

Molecular Location: base pairs 102,631,486 to 102,760,998 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- MANB1
- MANBA_HUMAN
- mannanase
- mannase
- mannosidase, beta A, lysosomal

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (1999): Glycoprotein Degradation
<https://www.ncbi.nlm.nih.gov/books/NBK20729/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28MANBA%5BTIAB%5D%29+OR+%28MANB1%5BTIAB%5D%29+OR+%28mannase%5BTIAB%5D%29+OR+%28mannanase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- MANNOSIDASE, BETA A, LYSOSOMAL
<http://omim.org/entry/609489>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_MANBA.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=MANBA%5Bgene%5D>
- HGNC Gene Family: Mannosidases type beta
<http://www.genenames.org/cgi-bin/genefamilies/set/1196>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6831
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/4126>
- UniProt
<http://www.uniprot.org/uniprot/O00462>

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